Muckle-Wells Syndrome (MWS): Fact Sheet

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What It Is

Muckle-Wells Syndrome (MWS) is a subtype of Cryopyrin-Associated Periodic Syndromes (CAPS). Diagnosis usually occurs shortly after birth.[1]

Causes and Symptoms

MWS is generally caused by mutations in a gene called *NLRP3* (nucleotide-binding domain, leucine-rich family [NLR] pyrin domain containing 3). *NLRP3* mutations cause increased activity of cryopyrin, a protein that regulates inflammation. Increased cryopyrin activity results in overproduction of a protein known as interleukin-1 beta (IL-1β).[1,2]

Overproduction of IL-1β, in turn, causes recurrent symptoms of inflammation such as[1]

- Rash
- Fever/chills
- Joint pain
- Eye redness/pain
- Fatigue

MWS symptoms are triggered by unknown, random factors and possibly by stress, exercise, or cold.[1,3] Episodes generally last 24 to 48 hours.[1] In most cases, patients with MWS develop progressive hearing loss.[4,5] In some cases, amyloidosis, a disease in which an abnormal accumulation of the protein amyloid occurs in tissues and organs, develops later in life.[3,5,6]

Living with MWS

Most patients with MWS experience chronic, daily symptoms of inflammation.[1] Chronic fatigue may severely impact these patients. Symptoms are often worse at night.[7]

Diagnosis and Treatment

Diagnosis of MWS is determined through an evaluation of a patient's symptoms and medical history. Confirmation of the diagnosis is sometimes achieved through genetic testing and the identification of *NLRP3* mutations.[8] Not all patients, however, have a detectable genetic mutation, making accurate symptom evaluation critical.[2]

There are FDA-approved therapies to treat the symptoms of MWS.[3]